

Plasmalogens (Red Blood Cells)

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Plasmalogens are a subclass of glycerophospholipids that play a critical role in maintaining the structural integrity and function of cellular and subcellular membranes. Their biosynthesis begins in peroxisomes; hence, peroxisomal biogenesis disorders (PBDs) such as Zellweger spectrum disorder (ZSD) can lead to significantly reduced plasmalogen levels. Moreover, markedly reduced plasmalogen levels are the hallmark of rhizomelic chondrodysplasia punctata (RCDP), a group of disorders caused by inherited defects in plasmalogen biosynthesis.

Featured ARUP Testing

[Plasmalogens \(Red Blood Cells\) 3019936](#)

Method: Quantitative Liquid Chromatography-Tandem Mass Spectrometry

Methodology

This assay uses liquid chromatography-tandem mass spectrometry (LC-MS/MS) to quantify the absolute concentrations, expressed in nmol/mL, of the eighteen most abundant ethanolamine plasmalogen species.¹ Results are reported as total values summing species based on the fatty alcohols at the plasmalogen *sn*-1 position.

Indications for Use

Plasmalogen testing is used in the diagnostic evaluation of inborn errors of metabolism that affect plasmalogen synthesis, such as ZSD or RCDP. It can also be used to monitor patients receiving therapies aimed at augmenting plasmalogen levels.

Test Interpretation

Plasmalogen levels are markedly reduced in patients with RCDP and ZSD. To account for their age-related physiological variation, age-specific reference intervals were validated to ensure accurate interpretation of assay results.

While more data is needed to clarify the relationship between plasmalogen levels and ZSD, a well-established correlation exists between the severity of plasmalogen deficiency and the clinical severity of RCDP.^{1,2,3}

Results

Total concentrations of the six 16:0, 18:0, and 18:1 plasmalogen species, as well as the overall total across all 18 plasmalogen species, are reported. Individual plasmalogen species concentrations are not reported.

Values below 60% of the age-specific control median are indicative of plasmalogen deficiency.

Limitations

- Plasmalogen testing may have limited diagnostic utility in older children with suspected PBDs, as plasmalogen levels may normalize with age.²
- Hemolyzed blood samples may lead to artificially low plasmalogen measurements, potentially affecting diagnostic accuracy.²
- Blood samples collected posttransfusion may yield inaccurate results, since transfused RBCs can persist in circulation for up to 120 days.²

References

1. De Biase I, Yuzyuk T, Cui W, et al. Quantitative analysis of ethanolamine plasmalogen species in red blood cells using liquid chromatography tandem mass spectrometry for diagnosing peroxisome biogenesis disorders. *Clin Chim Acta*. 2023;542:117295.
2. De Biase I, Tortorelli S, Kratz L, et al. Laboratory diagnosis of disorders of peroxisomal biogenesis and function: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2020;22(4):686-697.

3. Wegwerth PJ, White AL, Stoway SD, et al. [A new test method for biochemical analysis of plasmalogens in dried blood spots and erythrocytes from patients with peroxisomal disorders.](#) *J Inherit Metab Dis.* 2023;46(6):1159-1169.

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